

WHAT IS GENETIC COUNSELLING?

Many conditions or disorders tend to run in families. Genetic or hereditary conditions may be passed on from parents to their children, since each parent passes on half of their inherited material (also called genes) to a child. Some genetic conditions occur only once in a family. Genetic counsellors can give information about how these disorders are inherited and how likely they are to happen in a family.

Genetic counselling is a process whereby individuals and their families are assisted in addressing their concerns relating to the presence of or risk of a genetic disorder in themselves or a family member.

The health professional team providing genetic counselling may consist of clinical geneticists, other clinicians, genetic counsellors and genetic nurses.

The medical diagnosis, prognosis, the genetics and recurrence risks of the presenting disorder are dealt with at a genetic counselling consultation.

Genetic counselling also aims to provide families with options and support to deal with their risks and manage the disorder in the best possible way. The genetic counsellor will explore how the individual and family are coping with the condition and can refer them to appropriate support structures.

In many cases a genetic counsellor can reassure families that there is no particular risk or that the risk is much less than they feared. In some cases, there is a significant risk of recurrence and it is important that the family are made aware of their options for preventing recurrence in the family (including carrier testing and prenatal testing)

WHO SHOULD HAVE GENETIC COUNSELLING?

You may benefit from genetic counselling if:

- You or your partner has a condition which might be passed on to your children. Some examples of genetic disorders include: Cystic Fibrosis, Haemophilia A/B, Fragile X syndrome, Huntington Disease, Duchenne Muscular Dystrophy, Neurofibromatosis, Down syndrome or a chromosome abnormality.
- You or your partner has had a child or family member with a birth defect, developmental delay or a condition which may be genetic.
- You and your partner are related to each other (eg cousins) and intend to have children.

- You or your partner is a woman aged 35 years or older who is pregnant or planning a pregnancy.
- You or your partner has had three or more miscarriages.
- Ultrasound markers or fetal abnormalities have been detected in your pregnancy.
- You have had a positive screening test in pregnancy such as the triple test or nuchal translucency screening.
- You or your partner has been exposed to teratogens (eg infection, prescription or illicit drugs, alcohol) during a pregnancy.
- You or your partner has two or more relatives who have had cancer at a young age.
- You or your partner belongs to a particular ethnic group that is at risk for certain genetic diseases (eg Ashkenazi Jewish, Asian Indian).

WHAT HAPPENS AT A GENETIC COUNSELLING SESSION?

The genetic counsellor will discuss the client's reasons for seeking genetic counselling and any particular concerns.

The client will be asked for details of his/her family's medical history and a family tree (pedigree) will be drawn.

The medical professional will try to make or confirm a diagnosis and explain the management and implications of the genetic diagnosis. S/he will also explain the inheritance pattern of the condition to you.

The genetic counsellor will explain the management and implications of the genetic diagnosis and the inheritance pattern of the condition.

The risk that he/she may have a child with a particular condition can be estimated and explained.

The options for testing for the condition and the limitations of testing will also be discussed in detail if appropriate.

The genetic counsellor will discuss ways of coping with the condition and referral to other medical and social support systems available will be discussed. S/he will attempt to identify community resources that may be helpful.